1 This question is based on the case study ‘BRAF MUTATION TRIGGERS MOST MELANOMAS’ (Case Study 1).

You were told in the case study that in approximately 97 percent of BRAF gene mutations, the DNA sequence in malignant melanoma cells differed from that of healthy cells by just one base pair.

(a) Fig. 1.1 shows the structure of part of a DNA molecule.

(i) On Fig. 1.1 draw a box around a base pair.  

The answer to this question should be drawn on Fig. 1.1. [1]

(ii) Explain the importance of complementary base pairing in DNA.

..............................................................................................................................  
..............................................................................................................................  
..............................................................................................................................  
..............................................................................................................................  
..............................................................................................................................  
..............................................................................................................................  
..............................................................................................................................  
.............................................................................................................................. [2]
(b) Suggest why the BRAF gene is known as a proto-oncogene.

........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................[1]

(c) Fig. 1.2 shows the incidence of malignant melanoma in males and females, of different ages, in the United Kingdom (UK).

![Graph of incidence of malignant melanoma](image)

Fig. 1.2

(i) Using the information in Fig. 1.2, describe one similarity and one difference in the incidence of malignant melanoma in males and females.

........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................
........................................................................................................................................[3]
(ii) Suggest one reason for the similarity and one reason for the difference described in 1(c)(i).

similarity ...............................................................................................................................................
................................................................................................................................................................
................................................................................................................................................................
................................................................................................................................................................
difference ..................................................................................................................................................
.................................................................................................................................................................
.................................................................................................................................................................
.................................................................................................................................................................
................................................................................................................................................................. [2]

(d) Cell growth and cell division are controlled by cell signalling pathways.

Fig. 1.3 shows the BRAF protein and other proteins in a signalling pathway that normally controls cell division.

Fig. 1.3
(i) Fig. 1.3 shows that the signalling pathway is normally activated when growth factors bind to the cell surface membrane.

Suggest what feature of the cell surface membrane allows growth factors to bind.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
........................................................................................................................................... [2]

(ii) Describe the effect of the BRAF gene mutation on the signalling pathway.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
........................................................................................................................................... [2]

(e) You were told in Case Study 1 that the altered BRAF protein has a significantly different shape to the normal BRAF protein.

(i) Suggest why this difference in shape makes the altered BRAF protein an excellent candidate for drug targeting.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
........................................................................................................................................... [2]
(ii) The BRAF protein is an enzyme. Suggest how a drug might act to reduce or stop the activity of the altered BRAF enzyme. ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... [2]

(f) The research team carried out a phase 3 clinical trial to compare the effects of the drugs vemurafenib and dacarbazine, in treating malignant melanoma.

(i) Describe what is meant by a phase 3 clinical trial. ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... [2]

(ii) Most phase 3 clinical trials are randomised. Suggest how a randomised trial would be set up to compare the effects of vemurafenib and dacarbazine. ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... ........................................................................................................................................... [2]

[Total: 21]
This question is based on the case study ‘THE BURDEN OF CHRONIC DISEASE’ (Case Study 2).

(a) (i) Describe what is meant by the term *chronic* disease.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
........................................................................................................................................... [2]

(ii) State two chronic diseases other than chronic respiratory diseases and cancer.

1 ........................................................................................................................................
2 ........................................................................................................................................... [2]

(b) You were told in Case Study 2 that disability-adjusted life years (DALYs) can be used as an indicator of the overall health of a country.

Table 2.1 shows the age-standardised DALYs from chronic respiratory diseases, for men and women in India and the United Kingdom (UK), in 2004.

<table>
<thead>
<tr>
<th>Country</th>
<th>Gender</th>
<th>Age-standardised DALYs (per 100 000)</th>
</tr>
</thead>
<tbody>
<tr>
<td>India</td>
<td>female</td>
<td>1167</td>
</tr>
<tr>
<td></td>
<td>male</td>
<td>1387</td>
</tr>
<tr>
<td>United Kingdom</td>
<td>female</td>
<td>909</td>
</tr>
<tr>
<td></td>
<td>male</td>
<td>942</td>
</tr>
</tbody>
</table>

Table 2.1

(i) Explain why age-standardised figures were used.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
........................................................................................................................................... [2]
In India, males accounted for 54% of the total age-standardised DALYs.

In the UK, males accounted for a different percentage of the total age-standardised DALYs.

Using figures from Table 2.1, calculate this percentage.

Show your working and give your answer to the nearest whole number.

Answer = .................................................... % [2]

There are differences between India and the UK in the age-standardised DALYs resulting from chronic respiratory diseases.

Suggest explanations for these differences.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
........................................................................................................................................... [3]
Asthma is the most common chronic respiratory disease in children.

Fig. 2.1 shows a diagram of the cross-section through a normal bronchiole and a cross section through a bronchiole during an asthma attack.

Fig. 2.1

(i) With reference to Fig. 2.1, describe three changes that occur in bronchioles during an asthma attack.

1 ........................................................................................................................................................
........................................................................................................................................................

2 ........................................................................................................................................................
........................................................................................................................................................

3 ........................................................................................................................................................
........................................................................................................................................................

[3]
(ii) Two types of drug that are used to treat asthma are steroids and beta-agonists.

Describe how these drugs are introduced into the body and explain how they relieve the symptoms of asthma.

steroids ...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................

beta-agonists ................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................

(d) The Global Alliance against Chronic Respiratory Diseases (GARD) is trying to reduce the health burden of chronic respiratory diseases.

Suggest two policies that countries could introduce to prevent chronic respiratory diseases.

1 ................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................

2 ................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................

[Total: 21]
Karyotyping is a test which can help to identify genetic disorders.

(a) Fig. 3.1 shows a karyotype produced from a sample containing fetal cells.

(i) Name the genetic disorder shown in the karyotype in Fig. 3.1.

........................................................................................................................................... [1]

(ii) Describe how this karyotype differs from a normal karyotype.

........................................................................................................................................... [1]
(b) To check for genetic disorders in the fetus, karyotypes are made from cells isolated from a sample taken from the amniotic fluid or chorionic villi.

Describe how a karyotype is obtained from these cells.

...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
................................................................................................................................................... [5]
Genetic disorders, such as the one shown in the karyotype in Fig. 3.1, result from mistakes that occur when a gamete-producing cell divides by meiosis.

Fig. 3.2 below is a simplified diagram of meiosis in a gamete-producing cell. Only the X chromosomes are shown.

(i) On Fig. 3.2

- label the point where non-disjunction occurs
- label a diploid cell.

The answers to this question should be drawn on Fig. 3.2.
(ii) Explain how non-disjunction can lead to a fetus having the karyotype shown in Fig. 3.1.
Heart attacks are a major cause of death for both men and women worldwide.

Most heart attacks are caused by coronary heart disease (CHD).

(a) The following passage describes how CHD develops and leads to a heart attack.

Complete the passage by using the most appropriate terms.

In CHD, the lining of the coronary artery, known as the ......................................... becomes damaged and roughened. The ......................................... of the artery becomes narrower due to a build up of fatty deposits known as .......................................... A blood clot may form at this site and grow so large that it blocks the artery. This means that the cardiac ......................................... is no longer supplied with blood carrying ......................................... so aerobic respiration can no longer occur. This results in a heart attack. [5]

(b) State two symptoms of a heart attack.

...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
................................................................................................................................................... [2]

(c) A severe heart attack may lead to a cardiac arrest.

(i) Describe two signs that indicate that a person is having a cardiac arrest rather than a heart attack.

...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................
................................................................................................................................................... [2]

© OCR 2013
(ii) Cardiopulmonary resuscitation (CPR) is the first aid treatment for a person suffering from a cardiac arrest.

Describe the treatment given by a first aider, to a person identified as having a cardiac arrest, and describe what additional treatment may be given by a medically qualified practitioner.

*In your answer you should refer to both the treatment given by the first aider and the medical practitioner.*

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
........................................................................................................................................... [8]

[Total: 17]
Immunisation programmes have resulted in dramatic improvements in health. As a result of immunisation, several infectious diseases which were major causes of ill health are now rare in many countries.

Table 5.1 below shows the immunisation programme for young children in the United Kingdom (UK).

The vaccines used in the immunisation programme have code names, as shown in Table 5.1.

One of the vaccines and some of the diseases have been omitted from the table – these are indicated by the shaded areas.

<table>
<thead>
<tr>
<th>Age</th>
<th>Code name of vaccine</th>
<th>Diseases protected against</th>
</tr>
</thead>
<tbody>
<tr>
<td>2 months</td>
<td>DTaP/IPV/Hib (1st dose)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>PCV (1st dose)</td>
<td>pneumonia</td>
</tr>
<tr>
<td>3 months</td>
<td>DTaP/IPV/Hib (2nd dose)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>MenC (1st dose)</td>
<td>meningitis C</td>
</tr>
<tr>
<td>4 months</td>
<td>DTaP/IPV/Hib (3rd dose)</td>
<td></td>
</tr>
<tr>
<td></td>
<td>PCV (2nd dose)</td>
<td>pneumonia</td>
</tr>
<tr>
<td></td>
<td>MenC (2nd dose)</td>
<td>meningitis C</td>
</tr>
<tr>
<td>12–13 months</td>
<td>Hib (4th dose) / MenC (3rd dose)</td>
<td>measles, mumps and rubella</td>
</tr>
<tr>
<td></td>
<td>PCV (3rd dose)</td>
<td>pneumonia</td>
</tr>
<tr>
<td>3 years 4 months</td>
<td>DTaP/IPV/Hib (pre-school booster)</td>
<td>measles, mumps and rubella</td>
</tr>
</tbody>
</table>

Table 5.1
(a) (i) Using the information in Table 5.1, name the vaccine that protects children from measles, mumps and rubella.

........................................................................................................................................................................[1]

(ii) Name four of the diseases that children are protected against by having the DTaP/IPV/Hib vaccine given at 2, 3, 4 months and 3 years 4 months.

........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................[4]

(b) (i) Table 5.1 shows that young children need more than one dose of some vaccines.

Outline how a vaccine protects children from infectious disease and explain why more than one dose of the vaccine may be required to give full protection.

In your answer, you should make clear how the response to the second dose of the vaccine differs from the response to the first dose of the vaccine.

........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................
........................................................................................................................................................................

Answer lines for this question continue on the next page.
(ii) Suggest one reason why the routine vaccination programme in the UK starts when children are 2 months old.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
............................................................................................................................................... [9]

(iii) Suggest why some parents are worried about their children being vaccinated.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
............................................................................................................................................... [1]

(c) Sometimes a child’s immune system does not respond successfully to a vaccine.

In the condition known as PEM (protein-energy malnutrition), a child’s diet is low in fat and carbohydrates. This results in protein in the diet being used as an energy source.

Suggest why a vaccine may not produce an immune response in children with PEM.

...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
...........................................................................................................................................
............................................................................................................................................... [2]

[Total: 18]
Humans are multicellular organisms.

All the cells in the human body develop from a single fertilised egg which divides many times.

(a) Explain how cells become organised to form the **respiratory system** in the human body.
(b) Babies and infants need a balanced diet for healthy growth and development.

Complete Table 6.1 by inserting the name of a mineral or vitamin that is needed for each role in the body. Some minerals and vitamins have more than one role.

<table>
<thead>
<tr>
<th>Role in body</th>
<th>Mineral or vitamin</th>
</tr>
</thead>
<tbody>
<tr>
<td>for the functioning of the retina in the eye</td>
<td>vitamin A</td>
</tr>
<tr>
<td>to make haemoglobin</td>
<td></td>
</tr>
<tr>
<td>for blood to clot</td>
<td></td>
</tr>
<tr>
<td>for the development of strong bones</td>
<td></td>
</tr>
<tr>
<td>to make DNA, RNA and ATP</td>
<td></td>
</tr>
<tr>
<td>necessary for collagen and healthy skin</td>
<td></td>
</tr>
</tbody>
</table>

Table 6.1

(c) Babies and infants also need a source of essential fatty acids and essential amino acids.

State what is meant by the term *essential* in this context.

...................................................................................................................................................
..............................................................................................................................................
...................................................................................................................................................
...................................................................................................................................................

[Total: 10]

END OF QUESTION PAPER
If additional answer space is required, you should use the following lined pages. The question number(s) must be clearly shown in the margins.